DCX gene

doublecortin

Normal Function

The *DCX* gene provides instructions for producing a protein called doublecortin. This protein is involved in the movement of nerve cells (neurons) to their proper locations in the developing brain, a process called neuronal migration. Doublecortin attaches (binds) to microtubules, which are rigid, hollow fibers that make up the cell's structural framework (the cytoskeleton). The binding of doublecortin promotes the stability of microtubules. Microtubules help propel neurons by forming scaffolding within the cell that elongates in a specific direction, altering the cytoskeleton and moving the neuron.

Health Conditions Related to Genetic Changes

isolated lissencephaly sequence

More than 70 mutations in the *DCX* gene have been found to cause isolated lissencephaly sequence (ILS). This condition is characterized by abnormal brain development that results in the brain having a smooth appearance (lissencephaly) instead of its normal folds and grooves. Individuals with ILS have severe neurological problems, including intellectual disability and recurrent seizures (epilepsy) that begin in infancy. Most of the *DCX* gene mutations that cause ILS change a single protein building block (amino acid) in doublecortin and usually result in a protein with little or no function. A lack of normal doublecortin affects the stability and organization of microtubules, impairing their ability to move cells. Neurons in the developing brain are particularly affected, resulting in the neurological problems associated with ILS.

other disorders

Mutations in the *DCX* gene can cause a condition called subcortical band heterotopia, which is characterized by abnormal brain development, often less severe than ILS (described above). Subcortical band heterotopia occurs when neurons migrate to an area of the brain where they are not supposed to be (heterotopia), and form band-like clusters of white tissue. Since these bands are located beneath an area of the brain known as the cerebral cortex, they are said to be subcortical. The symptoms of subcortical band heterotopia depend on the severity of the brain abnormalities and can vary from severe intellectual disability and epilepsy to normal intelligence with mild or no epilepsy.

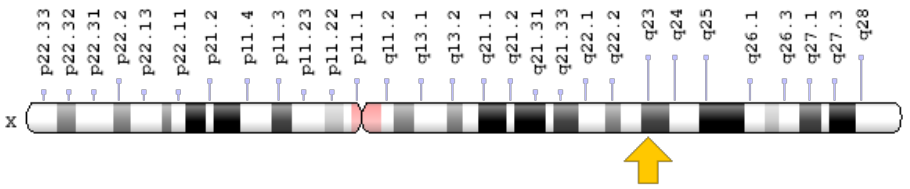
Subcortical band heterotopia usually affects females with a mutation affecting one copy of the *DCX* gene in each cell, while males with one *DCX* gene mutation

usually have ILS. Females can develop ILS and males can develop subcortical band heterotopia, but these instances are rare.

Chromosomal Location

Cytogenetic Location: Xq23, which is the long (q) arm of the X chromosome at position 23

Molecular Location: base pairs 111,293,779 to 111,412,232 on the X chromosome (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DBCN
- DC
- DCX_HUMAN
- doublecortex
- doublecortex; lissencephaly, X-linked (doublecortin)
- lissencephalin-X
- LISX
- SCLH
- XLIS

Additional Information & Resources

Educational Resources

- Neuroscience (second edition, 2001): Early Brain Development https://www.ncbi.nlm.nih.gov/books/NBK11113/
- Neuroscience (second edition, 2001): Neuronal Migration https://www.ncbi.nlm.nih.gov/books/NBK10831/

GeneReviews

- DCX-Related Disorders
  https://www.ncbi.nlm.nih.gov/books/NBK1185

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28DCX%5BTI%5D%29+OR+%28doublecortex%5BTIAB%5D%29+OR+%28X-linked+lissencephaly%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D

OMIM

- DOUBLECORTIN
  http://omim.org/entry/300121

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_DCX.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=DCX%5Bgene%5D
- HGNC Gene Family: Doublecortin superfamily
  http://www.genenames.org/cgi-bin/genefamilies/set/1369
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=2714
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/1641
- UniProt
  http://www.uniprot.org/uniprot/O43602

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